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Guanidinoacetate methyltransferase (GAMT) deficiency diagnosed by proton NMR spectroscopy of body fluids.

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Abstract

In patients with guanidinoacetate methyltransferase (GAMT) deficiency several parameters may point towards the diagnosis of GAMT deficiency. These include the low levels of creatine and creatinine in urine, the high concentration of guanidinoacetic acid (GAA) in urine and the low levels of creatine and creatinine in the cerebrospinal fluid (CSF). In this study, body fluids from 10 GAMT deficient patients were analysed using (1)H NMR spectroscopy. The urine 1D (1)H NMR spectra of all the patients showed a doublet resonance at 3.98 ppm (pH 2.50) derived from GAA present in high concentration. For this compound, a good recovery and good correlation was found between an LC-MS/MS method and (1)H NMR spectroscopy. In CSF NMR spectra of these patients, the singlet resonances of creatine and creatinine (3.05 and 3.13 ppm, respectively) were absent (normally always present in (1)H NMR spectra of CSF). Due to overlap by other resonances, the doublet of GAA could not be observed. Our data demonstrate that (1)H NMR spectroscopy of urine and CSF can be used to diagnose patients with GAMT deficiency.

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